



SETBP1 gene

SET binding protein 1

Normal Function

The *SETBP1* gene provides instructions for making a protein called SET binding protein 1 (SETBP1), which is found in cells throughout the body. The SETBP1 protein is known to attach (bind) to another protein called SET. However, the function of the SETBP1 protein and the effect of its binding to the SET protein are unknown.

Health Conditions Related to Genetic Changes

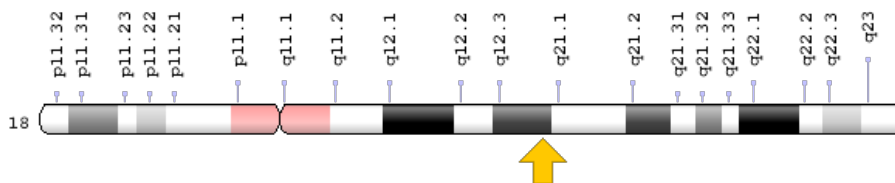
Schinzel-Giedion syndrome

At least five mutations in the *SETBP1* gene have been identified in children with Schinzel-Giedion syndrome, a severe condition apparent at birth that is characterized by distinctive facial features and abnormalities in many body systems. The gene mutations in affected individuals cluster in one region of the gene known as exon 4. However, the effects of the mutations on the function of the gene or the protein are unknown. Researchers are working to understand how mutations in the *SETBP1* gene cause the signs and symptoms of Schinzel-Giedion syndrome.

Chromosomal Location

Cytogenetic Location: 18q12.3, which is the long (q) arm of chromosome 18 at position 12.3

Molecular Location: base pairs 44,680,173 to 45,068,510 on chromosome 18 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KIAA0437
- SEB
- SET-binding protein
- SET-binding protein isoform a
- SET-binding protein isoform b
- SETBP_HUMAN

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SETBP1%5BTIAB%5D%29+OR+%28SET+binding+protein+1%5BTIAB%5D%29%29+OR+%28%28SEB%5BTIAB%5D%29+OR+%28SET-binding+protein%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

OMIM

- SET-BINDING PROTEIN 1
<http://omim.org/entry/611060>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/SETBP1ID44031ch18q12.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SETBP1%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=15573
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/26040>
- UniProt
<http://www.uniprot.org/uniprot/Q9Y6X0>

Sources for This Summary

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